LDHB gene

lactate dehydrogenase B

Normal Function

The *LDHB* gene provides instructions for making a protein called lactate dehydrogenase-B, which is one piece (subunit) of the lactate dehydrogenase enzyme. There are five different forms of this enzyme, each made up of four protein subunits. Various combinations of lactate dehydrogenase-B subunits and lactate dehydrogenase-A subunits (which are produced from a different gene) make up the different forms of the enzyme. The lactate dehydrogenase enzyme, which is found throughout the body, is important for creating energy for cells. The simple sugar glucose is the energy source for most cells. In the final step of glucose breakdown, most forms of the lactate dehydrogenase enzyme convert the molecule pyruvate into a similar molecule called lactate, which can be used by the body for energy. Other forms of this enzyme can convert lactate back to pyruvate, which can participate in other chemical reactions to create energy.

The version of lactate dehydrogenase made of four lactate dehydrogenase-B subunits is found primarily in heart (cardiac) muscle. This version of the enzyme converts lactate to pyruvate.

Health Conditions Related to Genetic Changes

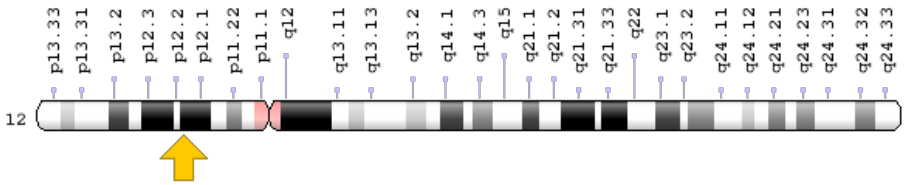
lactate dehydrogenase deficiency

More than 15 mutations in the *LDHB* gene have been found to cause lactate dehydrogenase deficiency, specifically lactate dehydrogenase-B deficiency. This condition is associated with a decrease in functional lactate dehydrogenase enzyme, but it does not appear to cause any physical signs or symptoms. Most *LDHB* gene mutations change single protein building blocks (amino acids) in the lactate dehydrogenase-B subunit. *LDHB* gene mutations lead to the production of an abnormal lactate dehydrogenase-B subunit that cannot form the lactate dehydrogenase enzyme. Even though lactate dehydrogenase activity is decreased in cardiac muscle cells of people with lactate dehydrogenase-B deficiency, they do not appear to have any signs or symptoms related to their condition. It is unclear why this type of enzyme deficiency does not cause any health problems.

Chromosomal Location

Cytogenetic Location: 12p12.1, which is the short (p) arm of chromosome 12 at position 12.1

Molecular Location: base pairs 21,635,342 to 21,657,971 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- L-lactate dehydrogenase B chain
- lactate dehydrogenase H chain
- LDH-B
- LDH-H
- LDH heart subunit
- LDHB_HUMAN
- LDHBD

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Isozymes of Lactate Dehydrogenase https://www.ncbi.nlm.nih.gov/books/NBK22392/figure/A1362/
- Biochemistry (fifth edition, 2002): Lactate and Alanine Formed by Contracting Muscle Are Used by Other Organs https://www.ncbi.nlm.nih.gov/books/NBK22423/#A2286
- Research Collaboratory for Structural Bioinformatics Protein Data Bank: Lactate Dehydrogenase http://pdb101.rcsb.org/motm/102

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28LDHB%5BTI%5D%29+OR+%28lactate+dehydrogenase+B%5BTI%5D%29+OR+%28lactate+dehydrogenase+H%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

OMIM

- LACTATE DEHYDROGENASE B
  http://omim.org/entry/150100

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_LDHB.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=LDHB%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6541
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/3945
- UniProt
  http://www.uniprot.org/uniprot/P07195

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- Maekawa M, Kanno T. Laboratory and clinical features of lactate dehydrogenase subunit deficiencies. Clin Chim Acta. 1989 Dec 15;185(3):299-308.
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- Okumura N, Terasawa F, Ueno I, Oki K, Yamauchi K, Hidaka H, Tozuka M, Okura M, Katsuyama T. Genetic analyses in homozygous and heterozygous variants of lactate dehydrogenase-B (H) subunit--LD-B Matsumoto I and II (LD-B W323R). Clin Chim Acta. 1999 Sep;287(1-2):163-71.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10509905
- Sudo K, Maekawa M, Kanno T, Li SS, Akizuki S, Magara T. Premature termination mutations in two patients with deficiency of lactate dehydrogenase H(B) subunit. Clin Chem. 1994 Aug;40(8):1567-70.
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